Hemoglobinopathy Fact Sheet

Hemoglobin E

*Hemoglobin** E is an inherited variant of normal adult hemoglobin (hemoglobin A). It results from a single change in one of the hundreds of *amino acids* that make up hemoglobin. Hemoglobin E is relatively common, particularly among people of Southeast Asian heritage (Cambodian, Laotian, Vietnamese, Thai). Other groups where it is found include Chinese, Filipino, Asiatic Indians and Turks. Summarized below are the three most commonly encountered hemoglobin patterns that involve hemoglobin E. *Genetic counseling* is advisable for families affected by these conditions to promote understanding of the significance for them or for future offspring.

Hemoglobin E Trait (*phenotype* FAE in infants and AE in adults)

Hemoglobin E trait results when a person inherits a hemoglobin E *gene* from one parent and a hemoglobin A gene from the other parent. This does not cause any health problems. For an infant identified with hemoglobin E trait on two newborn screening specimens, no further testing is indicated. However, other family members may be interested in having their individual phenotypes determined to learn if they have hemoglobin patterns involving hemoglobin E that can cause health problems for themselves or future children.

Homozygous Hemoglobin E (*phenotype* FE in infants and EE in adults)

Homozygous hemoglobin E results when a person inherits only hemoglobin E *genes*, one from each parent. A person with homozygous hemoglobin E has no genes for hemoglobin A. This condition is benign, but sometimes causes a mild anemia as shown by mild *microcytosis* on blood *indices*. For an infant identified with homozygous hemoglobin E by newborn screening, the findings should be confirmed by analysis of blood indices after two months of age, and determination of hemoglobin phenotype after nine months of age to rule out hemoglobin E-*beta thalassemia*, a more severe condition (see below).

Hemoglobin E-*Beta Thalassemia* (*phenotype* FE or FEA in infants and EF or EAF in adults)

Hemoglobin E-beta thalassemia results when a person inherits a hemoglobin E *gene* from one parent and a beta thalassemia gene from the other parent. This condition **has clinical significance**. Individuals with E-beta thalassemia will develop moderate to severe anemia. In some individuals the anemia is so severe that transfusions may be required. For an infant suspected of having E-beta thalassemia, the findings should be confirmed by analysis of blood *indices* after two months of age, and determination of hemoglobin phenotype after nine months of age. Both parents should also have a hematologic evaluation, if possible.

** This and all other italicized words are defined on the reverse side of this sheet.*

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DEFINITIONS of *italicized* words

Hemoglobin - the proteins in red blood cells that carry oxygen to body tissues.

Amino acids - building blocks of proteins.

Genetic counseling - meeting with someone knowledgeable in genetics to discuss information and risks about an inherited condition.

Phenotype - a characteristic produced by a set of genes (the specific set of genes is called the genotype).

Gene - a unit of inheritance that codes for a specific protein.

Microcytosis - small red blood cells.

Indices - set of values used to describe red blood cell characteristics.

Beta thalassemia - decreased production of one of the parts that make up hemoglobin.